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Translations of risk: decision making in a cancer genetics service

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Abstract *This paper reports on the routine decision making practices of health professionals working within a genetics service. Specifically the paper focuses on professionals' assessments of whether patients are at increased risk of inheriting a gene that predisposes the individual to developing cancer. By analysing professionals' talk and actions whilst assessing risk, the paper develops an understanding of both routine and complex decision making. Whilst some decisions were unproblematic, there were a number of situations in which geneticists appear to challenge the decision rules. Such situations occur when faced with a borderline case, an interesting case, when the patient appears particularly anxious or when the rules themselves are judged to be inadequate. Whilst decision support technologies are able to assist clinical decision making they are not relied upon by geneticists above and beyond their own knowledge. Thus, when knowledge is uncertain the decisions that clinicians make are not a consequence of the standard application of scientific protocols but are locally determined according to experience, circumstance and case. This suggests that the transfer of decision support technologies to general practice or to the general public are unlikely to be effective, given the importance of clinical experience to the risk assessment process.*

Key words: risk, cancer, genetics, decision making

Introduction

Recent advances in knowledge of the genetic basis of certain cancers have led to an increased awareness of the importance of a family history in determining susceptibility to the disease. Although increasing numbers of individuals are seeking advice from health care professionals in connection with their family history, the vast majority of cancer occurs sporadically with no clear familial pattern. The decision as to whether an individual is considered to be at high, moderate or low risk of inheriting a genetic predisposition to cancer can have important implications. Consequences of the determination of risk range from important treatment and screening related decisions to patients' psychological responses to that risk allocation. Whilst population based estimations of risk have mostly been the domain of epidemiologists, clinicians are faced with presentations of risk by individual patients. As such clinicians are

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required to identify risk as an object of diagnosis (Prior 2001) and devise management plans to address it.

Estimates of being at risk of a cancer are typically population based and expressed as an average lifetime risk, for example, currently around 1 in 12 for female breast cancer, 1 in 70 for ovarian cancer or 1 in 30 for colon cancer (Kerr *et al.* 2001). However, for any individual, risk may be either higher or lower than this average depending on a range of factors such as age, diet, exposure to environmental carcinogens or a family history of cancer. A number of mathematical models have been developed to assist with the estimation of cancer risk (Claus *et al.* 1994; Gail *et al.* 1989), however such models apply different weighting to various personal and biographical factors. The existence of different mathematical models are evidence in themselves of the competing versions of statistical probability within the calculation of genetic risk.

The development of evidence based medicine or protocols, which distil evidence from scientific research into recommendations for practitioners, has promoted a rational approach to medical decision making in recent years. The benefits of the evidence based medicine movement is considered to be the promotion of effective standardised treatments which are based on clear external evidence, untainted by the local contingencies of practice. Part of this movement includes the development of protocols or guidelines across many clinical specialities that serve to standardise referral and treatment decisions. Flynn (2002) has argued that medicine combines aspects of 'embrained knowledge' (individual, formal knowledge dependent on cognitive skills) and 'embodied knowledge' (individual, tacit, practical and context specific), but that clinical governance represents a drive to transform medicine into 'encoded knowledge' (collective, explicit and codified) through the implementation of clinical guidelines. However, whilst it is important to base treatment decisions on effective evidence, it should be remembered that health professionals negotiate problems and decision-making in what are often ambiguous or uncertain circumstances (Bloor 1976; Pope 2002; White 2002). Furthermore clinical genetics is unusual in that there is often no clear external evidence, due to it being a new and developing speciality. This paper is concerned with how professionals accomplish decision making in practice alongside decision support tools such as referral guidelines and risk estimation technology. In so doing a more sophisticated understanding of the complexities of professional decision-making surrounding risk estimation is presented.

Background to the cancer genetics service

The cancer genetics service that lies at the core of this study receives approximately 30 referrals per week from primary and secondary care. The service is required to quickly triage referrals to identify patients who may be at an increased risk of inheriting a familial cancer, and could therefore benefit from further investigation. With input from lead cancer specialities and primary care colleagues, the service has developed a set of rules (or guidelines) in order to distinguish which individuals or families, are at increased risk (see Figure 1). The guidelines, introduced in July 2000, have been distributed to secondary care specialists and general practitioners to help them assess the relative risks for their patients and avoid unnecessary referral. The estimation of cancer risk is important as the level of risk inevitably effects management of the patient. Patients that fulfil the guidelines are deemed to be at either moderate or high risk and would be accepted into the cancer genetics service for further investigation of their family history of cancer. Such patients would consequently receive expert assessment and advice about their cancer risk. Access to additional or early screening (such as mammography or colonoscopy) may be recommended. For some high-risk individuals genetic mutation testing may be offered following a series of clinical counselling sessions. An important element of the work of a cancer genetics team is to make decisions about which patients are at most risk. Directing services to individuals at increased risk improves the

We will accept referral where the following criteria are met:

Breast Cancer
- One first degree relative diagnosed at 40 years or less
- 2 first degree relatives at 60 years or less (on the same side of the family)
- 3 first or second degree relatives any age (on the same side of the family)
- 1 first degree male breast cancer
- A first degree relative **with bilateral breast cancer**

Breast / Ovarian Cancer
- Minimum: 1 of each cancer in first degree relatives (if only one of each cancer, the breast cancer diagnosed under 50 years)
- A first degree relative who has **both** breast and ovarian cancer

Ovarian Cancer
- 2 or more ovarian cancers, at least one first degree relative affected (on the same side of the family)

Colon Cancer
- 1 first degree relative diagnosed at age 40 or less
- 2 first degree relatives at 60 years or less (on the same side of the family)
- 3 relatives, all on the same side of the family, (at least 1 should be a first degree relative)
- Familial Adenomatous Polyposis
- Hereditary non polyposis colorectal cancer (revised Amsterdam criteria)

Other Cancer Syndromes
- Patient from a family with a known single gene cancer syndrome: von Hippel-Lindau disease, multiple endocrine neoplasia, retinoblastoma
- "Related Cancers": There are some rare cancer syndromes (e.g. Li Fraumeni syndrome and Cowden syndromes) where a variety of different cancers occur within a family. Where there is a high index of suspicion, the possibilty of referral should be discussed on an individual basis.

Figure 1. *The referral guidelines for individuals with a family history of cancer*

effectiveness of health interventions by targeting patient populations where research evidence has demonstrated that they may benefit from those interventions. In addition it is hoped that patients deemed not at increased risk will be spared unnecessary anxiety by keeping them out of secondary care and promoting reassurance from the GP.

Data collection

The data presented in this paper were collected during an Economic and Social Research Council (ESRC) funded ethnographic study that aims to examine the manner in which technologies of risk assessment are operationalised within a cancer genetics service. As such the research aimed to provide a detailed case–study of how 'expert' (scientific, medical and nursing) knowledge, technological artefacts and social processes intertwine in the routine work of risk assessment. Fieldwork was carried out in a regional cancer genetics service based within a large hospital in the UK. The cancer genetics service is made up of a number of professionals working together including clinical geneticists, genetic nurse specialists and laboratory scientists. Staff within the service may discuss a patient's case in a number of contexts, but primarily during their regular meetings. It was in such meetings that the 'naturally occurring data' (that is, data that would exist independently of the researcher's presence), were collected.

Data were gathered by means of audio-tape recordings and note taking (undertaken by the first named author). In total 25 meetings were audio-recorded over a period of 8 months. The study was approved by the relevant Research Ethics Committees and undertaken with the full knowledge and consent of all the participating staff.

Ethnographic research emphasises the need to think oneself into the perspective of the members of social group that one is studying. Perhaps the best way for the ethnographer to achieve this is to become part of the 'natural setting'. Observation is considered to be a key instrument in acquiring ethnographic knowledge. In particular observation can be usefully applied in studies of professional decision making as the technique is able to provide data on routine activities and reveals insights into non-rational or subjective decision making processes that may be less likely to be disclosed in an interview (McKeganey *et al.* 1988). Observational work can therefore expose the more chaotic and situational character of decision making.

Our data were collected from two types of meeting. The first of these were the referral meetings (13 meetings), and the second were risk review meetings (12 meetings). The referral meeting is a weekly event usually attended by one or more consultant clinical geneticists, a number of genetic nurse specialists, administrative staff and sometimes laboratory scientists. The meetings took the format of either the consultant or the senior nurse reading aloud the referral letter, usually verbatim, but sometimes summarising the salient points of longer letters. Following the reading the referral letters were then allocated to one of two main outcomes. Either the referral was accepted into the service for further investigation of the referred patient's family history or the referral was not accepted on the grounds that the patient's family history, as described in the referral letter, did not present a significant family history of cancer. If the referral were accepted, a family history questionnaire would be subsequently sent to the patient in order to collect further details about the medical history of the patient's relatives. If the referral were not accepted, the consultant would then write back to the referring doctor either requesting further details about the family history or briefly explaining that the patient's family history was not currently significant but that they should re-refer the patient if circumstances were to change. Figure 2 presents the geneticists' system of acceptance by means of a decision tree, which as Gladwin (1989) argues, can be modelled through the collection of ethnographic data.

Decision making as pattern recognition

In many cases the decision of whether or not to accept the referral was swift, and accomplished by the consultant reading the referral letter with no discussion required or offered from others present. Such unproblematic cases were usually the consequence of the family history clearly complying with the referral guidelines. The routine, unproblematic referrals demonstrate how clinical decision making can, in its simplest form, resemble a form of pattern recognition and response. Other sociological studies of clinical work have also demonstrated that, as in other aspects of life, the rules which give activity its meaning are often not articulated. For example Bloor (1976) gathered observational data in ENT clinics in order to explore the routines used within everyday clinical practice. Drawing on a phenomenological perspective (Schutz 1967), Bloor explained that the process of interpreting stimuli and acting upon them can be extremely elaborate, entailing a number of subsidiary projects to be embarked upon or, on the other hand, they may be swift due to its simplicity or the frequency of its occurrence. In the latter situation, everyday action is effortlessly followed in a 'natural attitude' (Schutz 1967). As data extract 1 illustrates, during the initial stages of risk assessment, clinicians may engage in a non-verbalised process of pattern recognition that matches the referred family history to the guidelines. The clinician's action of writing on the referral letter is a legal requirement in that it provides an audit-trail of clinical decision making, but it also serves an immediate purpose in that it indicates to all present that the decision is made to accept the patient into the service.

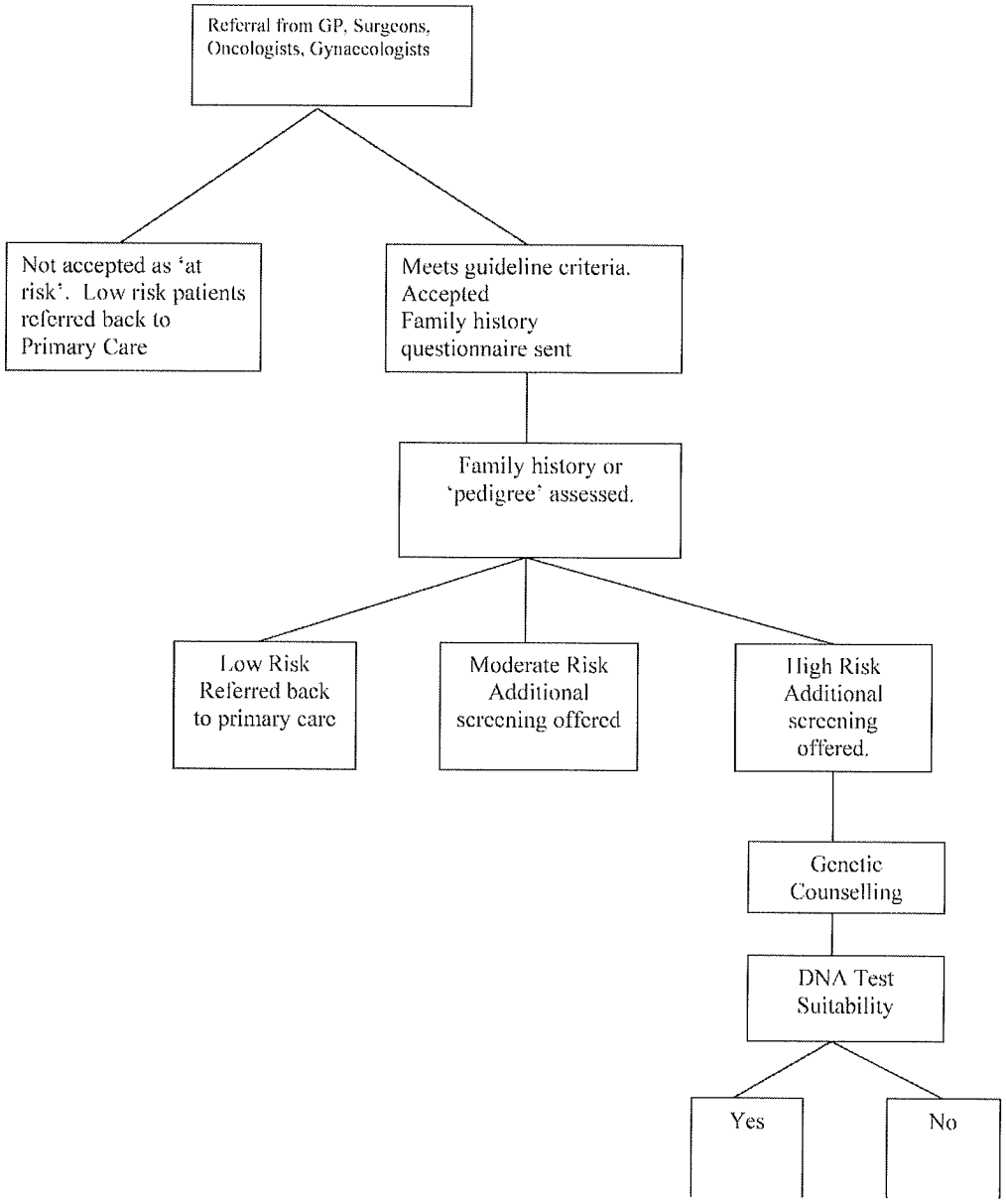

Figure 2. *Cancer geneticists' decision tree*

Furthermore this process remains unchallenged by others present as they all share a common understanding of the rules and the process of pattern recognition and response. A glossary of the transcription symbols used within all data extracts is provided at the end of this paper.

Extract 1.

1. CG1: ((Reads letter from a breast surgeon)) '*36-year-old lady whom I have*
2. *recently seen at the breast clinic with benign nodularity and I have discharged*
3. *her from this point of view. Her sister developed breast cancer at the age of*
4. *38 and she is understandably very concerned. I would be grateful if you*

5. *would give further advice.'* Fine, no problems. ((CG1 writes 'questionnaire'
6. on the referral letter, signs and dates it)).

Referral Meeting – 6.8.01

Dealing with borderline and ambiguous cases

On other occasions the decision about the outcome of the referral was more problematic and was accompanied by either a short or sometimes prolonged discussion by those present. The most common reason for this was that the referral was considered to be what the geneticists themselves referred to as a 'borderline case'. A borderline case could be the result of the exact ages of affected relatives not being explicitly stated in the referral letter, or the site of the cancer being unclear. In such instances cancer geneticists usually give the referral the benefit of the doubt. They justified their decision to allow these patients through on the basis that it is better to seek clarification about the family history and then confirm that the pedigree (family tree) is 'low' risk than to apply the guidelines rigidly and not follow up pedigree details. In the latter situation the team could potentially deny a higher risk individual a more in-depth assessment of their risk. In the following example the ambiguous nature of the patient's family history is evident at lines 5 and 7 with the repeated use of the word 'unsure' within the referral letter. In line 8 the clinical geneticist signals his intention to be generous given 'they only just don't fit' and the suggestion to accept the patient is endorsed by the genetics nurse at line 9 on the basis that the referring physician has endeavoured to provide as much information as possible.

Extract 2.

1. CG1: This is about ((patient name)). '*In reply to your letter about ((patient*
2. *name)) her family history of cancer is as follows. Sister was aged 62 and was*
3. *diagnosed with carcinoma of the colon in October of last year. Brother also*
4. *recently diagnosed with bowel cancer aged 63. (0.2) She tells me that her*
5. *father died aged 59 of cancer but is unsure of the site of origin. I'm not sure*
6. *whether this meets your referral criteria. She does have three relatives all*
7. *with cancer but they are unsure whether her father had colon cancer or not.'*
8. *(0.4)* So I'm tempted to send a questionnaire. They only just don't fit.
9. GN1: He has tried to give us as much information as possible, hasn't he?
10. CG1: Yes. ((CG1 writes 'questionnaire' on referral letter, signs and dates it)).

Referral Meeting - 3.12.01

Over-riding the guidelines

Despite the existence of the criteria to guide the referral and acceptance of patients, it became evident that during referral meetings the guidelines were not uniformly applied but were over-ridden by clinicians for a number of reasons. Patient anxiety was one reason why the team accepted patients into the service who did not fulfil the referral criteria. Typically such cases were characterised by a longer referral letter in which the referring physician constructed a more detailed picture of the emotional and medical history of the patient. Although details remained essentially factual, implicit in such constructions were persuasive accounts of the patient's anxiety and the referring physician's expectations for assistance from the genetics service. The genetics service response to such cases was invariably compassionate. In extract 3 the patient's desire to pursue prophylactic surgery to reduce their risk is interpreted as a significant indicator of patient anxiety and possibly an action that can be avoided if the patient's risk is deemed to be low. A more detailed examination of the family history and counselling of the patient by the specialist genetics service could serve to reassure the patient. Although the patient does not, on

empirical grounds, meet the referral guidelines the team wishes to accept the referral and consequently construct, and rehearse in front of each other, justifications for their decision. For example, at lines 13–15, CG1 lists the family cancers mentioned in the referral letter. The rhetorical function of recycling this information may be to ensure that all the cancers are taken into account. The patient's desire for surgery is mentioned again by GN1 at line 16 to emphasise that their decision about this patient has important consequences. Perhaps more significantly at lines 17 and 18 GN1 and GN2 discuss the possibility that the bone cancer could be a secondary cancer following metastatic spread from a primary breast cancer, although there is no evidence nor even a suggestion of this within the referral letter. However, if this were true then the patient would have a second relative with breast cancer and consequently a more obvious pattern of cancer inheritance would be evident. It does not take long before a decision is made to accept the referral and others present endorse the decision through their agreements that the patient does indeed sound very anxious (lines 22, 23).

Extract 3.

1. CG1: ((reading GP referral letter)) '*I would be grateful if you could see this*
2. *lady who is very concerned about the high incidence of cancer in her family*
3. *(0.4). She has not had any malignancy but does have repeatedly abnormal*
4. *smears and had several colposcopies including a laserdiathermy with a*
5. *GCIN2 (1.0). She has also requested a mammogram for reassurance (0.4)*
6. *although she has not felt any lumps and on examination has no lumps. In*
7. *terms of family she has a maternal aunt who has, as she terms it, bone cancer,*
8. *a maternal uncle with renal carcinoma. Her mother had breast cancer and*
9. *her grandmother had bowel cancer (0.2). All of these relatives were over 60*
10. *when these cancers were diagnosed. (0.4) She is really quite concerned and is*
11. *considering having a hysterectomy to prevent her developing cervical or*
12. *endometrial can:cer. (0.6) Previous medical history is otherwise*
13. *unremark:able.*' (1.0) So she has got a maternal aunt (0.2) with bone cancer, a
14. maternal uncle with renal cancer, mother with breast cancer, grandmother
15. bowel cancer.
16. GN1: It's difficult isn't it because if she is contemplating surgery
17. GN2: The bone cancer could be =
18. GN1: = Yes it could have been secondary breast couldn't it. (2.0)
19. CG1: Right I would be tempted to send her a questionnaire =
20. GN1: = Yeah
21. CG1: So at least she sees that we are taking things seriously.
22. GN1: She sounds an anxious] lady.
23. CG3: She sounds very anxious, doesn't she.
24. CG1: ((writes 'questionnaire' on the referral letter, signs and dates it))

Referral Meeting 3.12.01

Another observed reason for over-riding the referral guidelines was that the family history of the referred patient presented a significant, interesting or unusual case that the team felt deserved further investigation. In extract 4, a patient has been referred with a very strong family history of throat cancer which, as indicated in the referral letter, concerns the patient particularly in terms of his children's susceptibility (lines 5–7). There is currently no evidence for a genetic involvement in the aetiology of throat cancer and throat cancer is not mentioned on the referral guidelines as a cancer that requires genetic investigation. As the referral does not comply with the guidelines, it could, in a strict sense, be considered to be 'inappropriate'. The referring physician appears aware of this problem as she draws attention to it in her

referral (lines 7–10). However it is clear that the geneticists find the family history significant and indeed are surprised by such a high incidence of throat cancer within the family (lines 11 and 19). Although they acknowledge that the cause is more likely to be due to behaviour—as evidenced by the discussion of tobacco use (lines 20, 21)—they make the decision to accept the patient for further investigation. So why have the geneticists accepted the referral? One explanation that is suggested in the data at lines 22–26 is that the geneticists find the case genuinely interesting and are keen to investigate the circumstances surrounding the high incidence of throat cancer in the family. However they are also accepting the referral as they feel a duty of care to a concerned individual with such a significant family history of cancer. Whilst they know that the cause of the family's cancer is not genetic, the patient does not know this and consequently requires reassurance from a genetics service. As Bosk (1992) indicates, geneticists are required to attend to the emotional and psychological aspects of their clients problems and, in doing so, they face moral dilemmas in the course of their work.

Extract 4.

1. CG1: ((reading referral letter from GP)).... '*49-year-old. He appears to have*
2. *a very strong family history of throat cancer. I understand that his father died*
3. *from this in his 60s and his father's two sisters also died, one at 47 and the*
4. *other in her 60s. I also understand that both of his father's parents died in*
5. *their 50s with throat cancer. (2.0) Given this history he is concerned whether*
6. *there is something in the family that should be investigated. He tells me that*
7. *he is more concerned for his children than himself. I am unclear from your*
8. *guidelines* ((laughs)) *whether throat cancer is something that does have a*
9. *strong genetic component (0.4) and whether investigation of this patient is*
10. *warranted.*'
11. GN1: Oh, five family members! (1.0)
12. CG1: Right, father in his 60s
13. GN1: Yeah
14. CG1: Two of his =
15. GN1: Sisters
16. CG1: = sisters, one at [47
17. GN1: And both] parents
18. CG1: And both of his father's parents died with throat cancer.
19. GN1: Gosh! Quite a lot isn't it.
20. CG1: Perhaps they chewed tobacco.
21. GN3: Chewed tobacco, yeah, smoking, drinking
22. CG1: Are you happy to accept that as a sort of unusual interesting (0.8)—
23. nothing about breasts in there at all—sort of BRCA2? ((laughter))
24. CG2: I wouldn't presume for you on this one. I mean it's how you feel on this
25. one.
26. GN1: It's quite interesting.

Referral Meeting 23.7.01

Changing the rules to fit a case

Infrequently the team would suggest that their own referral guidelines should be reviewed. On such occasions a referral may have been received in which the patient's family history appeared significant as judged by the geneticists own experience or clinical judgement, but did not fulfil the guidelines as they stood at the time. In the following example the team discusses whether or not their guidelines on ovarian cancer are too strict and should be made more inclusive.

They compare the ovarian guidelines with the breast guidelines that they deem to be more generous. Although, during this meeting at least, they are not able to anchor their decision in firm evidence by pin-pointing exact published studies, they suggest that there is a case to be made for lowering the ovarian criteria. In doing so one of the cancer geneticists refers to his knowledge of epidemiological data (lines 11–13) which he considers provides justification to his opinion. Furthermore at lines 15–17 GN1 refers to an eminent consultant gynaecologist's technique of assessing ovarian risk which supports their feeling that their own guidelines are too strict. The decision is then made to accept the referral and the repeated endorsement of the decision at lines 15, 18, 22, 26 and 27 are perhaps an indication of teamwork in action. Although at this meeting they do not agree to amend their guidelines accordingly, the possibility of future amendment is flagged up, demonstrating that the guidelines are not viewed so much a set of universal rules but rather dynamic subject to ongoing validation and hence always under construction.

Extract 5.

1. CG1: ((reading GP referral letter)) '*Would you please see this 31-year-old*
2. *whose mother died of ovarian cancer at the age of 35 who would like to*
3. *discuss the implications. I am aware that your clinical guidelines state that*
4. *you will accept a referral for two or more ovarian cancers with at least one*
5. *first degree relative affected, (0.2) and in view of her mother's young age at*
6. *diagnosis ((patient name)) is naturally concerned.*' (2.0) I had one similar to
7. this that phoned me up. My feeling is that... basically we've got guidelines
8. for ovarian cancer, but we put it higher than all of the guidelines in relation to
9. breast cancer. But if breast cancer occurs at a young age of under 40 then we
10. would see them, and we put them at moderate risk. But we don't do that for
11. ovarian. And yet it doesn't make sense. Ovarian cancer carries a potentially
12. higher risk in relation to its weighting for BRCA. So I think if we see
13. someone with ovarian under 40 then we ought to regard them as the same way
14. as a breast.
15. GN1: Yes. I would agree. I mean ((gynaecologist name)) has this
16. back of the cigarette packet calculation that she deducts 10 years off the age of
17. diagnosis for ovarian, and that equates to a breast cancer age.
18. CG1: Yep, yep.
19. GN1: So if you have got somebody with a 40 ovarian then its 30 breast is the
20. equivalent risk. I'm not sure where she has got the evidence for that from. I'm
21. sure she is right but I don't know where the evidence is.
22. CG3: If you had a woman at 30 with breast you wouldn't need any other
23. relatives.
24. GN1: No. Just 1 under 40. But certainly ovarian because (0.2) it is more
25. likely to be hereditary with a young age.
26. CG1: With a young age, yep.
27. GN1: Yes I agree.
28. CG1: ((Writes questionnaire on the referral letter, signs and dates it.))

Referral Meeting 12.11.01

Producing risk assessments

Another site where patients' risk status is assessed is in the Risk Review meetings. Again these meetings are usually held weekly or fortnightly within the Cancer Genetics Service and are attended by consultant clinical geneticists, genetic nurse specialists, registrars and occasionally

students training in genetics. The purpose of the meeting is for genetic nurse specialists to present pedigrees (family trees) which they had 'worked up', to discuss any problems that have arisen during their investigations and to agree a risk classification for the referred patient. The printed pedigrees thus become a focus for the discussion and serve to both mediate and structure the meetings and the social interaction within it. The 'worked up' risk assessments are constructed through complex processes. Information is primarily supplied by patients who, through the medium of the family history questionnaire, provide details of both living and dead relatives. This information is often checked against other sources such as information provided by other family members, pathology reports, death certificates, hospital notes and cancer registry details. However, confirming cancer morbidity and mortality is only done on families who appeared to be at high risk of carrying a cancer gene or where the confirmation is deemed to be important. For example reports of familial ovarian cancer will be confirmed to establish whether the cancer was indeed ovarian or perhaps some other abdominal or uterine cancer. Another reason for confirmation, hinted at in extract 3, would be to establish whether the affected organ was a primary cancer site or due to secondary spread. Furthermore it is not always possible to confirm cancers as diagnosis may pre-date the cancer registry data or referred patients may have lost contact with relatives thus limiting the accuracy of their reports. Often genetic nurse specialists would have phoned patients in order to clarify missing or ambiguous details. Consequently family history information presented at risk assessment meetings is much more comprehensive than the brief details that the geneticists have at their disposal in the referral meetings.

Applying technology to assist the determination of risk

The translation of the detective work described above into a risk assessment is achieved through the production of a pictorial presentation of a family history (or pedigree). The representation of risk is achieved through a computer software tool known as Cyrillic and, as Fujimura (1996) has indicated, tools such as these are central to the manufacture of scientific data. Cyrillic is used by geneticists to draw a family tree, thus providing an explicit trace of hereditary influences, and to provide a numeric estimate of an individual's risk (Prior *et al.* 2002). Cyrillic is a classical example of an 'inscription device' (Latour and Woolgar 1986), that is, technological equipment that provides a visible trace of an invisible entity, thus turning it into documentary form so that it can be known and shown to others through its paper representation.

Scientific and medical practice are full of inscription devices which have been 'black boxed' (Latour 1987), that is, already successfully passed the point of being accepted into scientific practice and consequently deemed unproblematic. However, the programme is dependent on a statistical model that uses epidemiological data to inform its numerical calculations of risk. As described earlier, the role of family history amongst other variables may differ between models and consequently different risk estimates will be produced.

Over-riding guidelines and technology: the importance of clinical experience

Cancer geneticists often used Cyrillic's numerical calculations of risk to determine a patient's level of risk and allocate them to a 'risk group'. For example they operate a general rule that the cut-off between low and moderate risk is a lifetime risk of 15% and the cut-off between the moderate and high risk groups is a heterozygote risk of 25% (a heterozygote risk is the risk of the individual carrying a cancer predisposing gene). However, cancer geneticists sometimes preferred to use their own knowledge of genetic inheritance to over-ride the Cyrillic estimation, and clinical judgement was sometimes employed by geneticists to determine the

extent to which the recommendation of the decision support device were adhered to. This echoes Freidson's (1970) characterisation of the 'clinical mentality' as one in which the clinician values his/her own accumulation of personal first hand experience in preference to abstract principles or 'book knowledge', and which is a consequence of a liberal approach to practitioner autonomy and the exercise of clinical judgement. Physicians' reliance on personal experience at the expense of the uniform application of standard knowledge has also been exemplified by other sociological accounts of clinical decision making (Atkinson 1995; Berg 1997; Bloor 1976).

Geneticists may use their clinical judgement to either over-ride an estimate calculated by Cyrillic or use Cyrillic to justify a decision that their clinical experience suggests they should make. It is clear that Cyrillic is not the ultimate arbitrator of the decision but rather recruited selectively by the geneticists in order to firm-up their decision making.

In the following example the genetic nurse specialist presents the patient's case. In doing so the geneticists focus on the pictorial presentation of risk (Prior *et al.* 2002) which is imaged in the family tree or pedigree to which they point as they discuss the case (line 4). Although the referral letter states that the patient's mother had breast cancer, it is only when the patient returned the family history questionnaire and the cancer was confirmed with the Cancer Registry that the mother was found to have ovarian cancer at 55. The referred patient's mother also had a type of skin cancer (BCC or basal cell carcinoma) on her eyelid. In addition the referred patient's great grandmother had Hodgkin's lymphoma (a malignant disease of the lymphatic tissue). Neither the basal cell carcinoma nor Hodgkin's lymphoma have any genetic basis so the only remaining potentially familial cancer is the ovarian cancer. At lines 15–20 the clinicians express their surprise that Cyrillic has calculated the patient's risk to be moderate on just the basis of one ovarian cancer, one Hodgkin's disease and a further abdominal cancer (although it is unclear from the data extract which relative has this further abdominal cancer). In lines 21–23 they briefly discuss whether a reason for Cyrillic's surprisingly high calculation could be due to the relatively young age of onset of the ovarian cancer. However, it does not take long before the Cyrillic estimation is rejected in favour of the clinicians' own intuitive view that the patient is at low risk of ovarian cancer.

Extract 6.

1. GN2: OK, this one, the referral said '*mother recently died of breast cancer at*
2. *the age of 55. She also thinks that her great grandmother had the same*
3. *problem.'* (3.0)
4. CG1: So this is this lady up here? ((points to pedigree))
5. GN2: Yep, but in fact on her questionnaire she talked about abdo
6. ((abdominal)) area for mother. (2.0) This lady ((great grandmother)) was also
7. query an abdo but it came back as a Hodgkin's from registry. (1.0) So the
8. confirmed ovarian is her mum at 55. And she also had a BCC of her eyelid.
9. (0.4) Em, (0.4) and the skin that she talks about in that side ((paternal side))—
10. I know that that side is not relevant—but she talks about it being the same as
11. her mum. So BCC end of the market.
12. GN1: So you have got what? An ovarian and a =
13. GN2: = Hodgkins.
14. GN1: Hodgkins. (1.0) And an abdo.
15. CG1: Which I have to admit I would put at low risk.
16. GN1: Mm
17. CG1: But I am very interested in the heterozygote risk of 3.1% and an overall
18. risk of 16.5%, which puts her into a moderate risk. Based on what?
19. GN1: Mm, this is where it =

20. CG1: = Based on one ovarian cancer? (2.0)
21. GN2: ((sighs)) Maybe it's the age (1.0) 55. (1.0)
22. CG1: Mm
23. GN2: The younger end of the market.
24. CG1: OK. Ignore it. Low.
25. GN2: Low, thank you ((writes in patient's notes))

Risk Review Meeting – 1.10.01

Discussion

Previous research on lay and professional constructions of genetic risk have focused on the fundamental differences between biological and social understandings of inheritance and how these may be brought together (Atkinson *et al.* 2001; Parsons and Atkinson 1992; Richards 1996). The work of clinical geneticists involves dissecting the complexities of both social and biological relationships and translating knowledge of population risk to the individual case presented before them. As we have demonstrated this translation carries tensions due the ambiguities and uncertainties inherent within each patient's case and with the availability of evidence on epidemiological data. The link between population data and an individual case can only be forged through the decision making process.

Common to many decision-making models is a process of assembling facts and opinions and evaluating them in order to inform the decision (Allison 1969; McGrew and Wilson 1982). In many ways, therefore, cancer geneticists can be conceptualised as practical reasoners who, during the process of investigating a family history, assemble facts and make rational decisions on the basis of the evidence in front of them. This process of evaluation requires professional judgement. We have seen in this paper that decisions surrounding the allocation of an individual to high, moderate or low risk are based on judgements that are composed of a number of things. Thus, during their meetings cancer geneticists and genetic nurse specialists drew on a variety of information which they fed into their decision making process. Primarily this information was taken from the patient's pedigree as specified in the referral letter or as constructed from the family history questionnaire. In addition geneticists also drew on other information contained within the referral letter or questionnaire, for example evidence or suggestions of patient anxiety either from the referrer or from the patient or evidence of other physical health circumstances of the patient. Later on in the risk allocation process geneticists may try to confirm cancers perhaps through accessing hospital records, death certificates or by request to the local Cancer Registry. Geneticists also routinely draw on other information to assist their decision making. These can include research literature, other guidelines, opinions of other professionals working within their own or related specialities and their own professional experiences.

The referral guidelines were an important reference point in the decision making process and were frequently appealed to by the geneticists to justify either 'appropriate' or 'inappropriate referrals'. But the referral guidelines were not designed to serve as an inflexible set of rules and nor were they considered as such by the geneticists. Indeed the guidelines were modified by the team for a number of reasons: for borderline cases, for anxious patients, for significant and interesting cases, and where the rules themselves seemed inadequate. The data collected in this study also indicate that although decision support technologies such as Cyrillic are referred to during deliberations of risk categories, clinicians do not trust in these technologies and the resulting estimates above their own judgements. Clinical experience was the ultimate arbitrator of decisions about risk. This finding has important consequences for the management of patients. National data indicate an increasing number of referrals for people at low risk of inherited cancer to genetics clinics, which in turn has led to suggestions that general

practitioners need new skills to act as effective gatekeepers to over-stretched secondary services (Kinmouth *et al.* 1998). Consequently there has been speculation about the role that general practitioners can play in assessing and counselling patients of their risk (Emery *et al.* 1999), and following this the feasibility of using computer based systems to assess genetic risk within primary care (Emery *et al.* 2000). However if, as our data suggest, the experts in genetic risk assessment rely on confirming family histories and on their personal or tacit knowledge to estimate an individual risk, then transferring technologies from secondary to primary care is unlikely to lead to a transfer of expertise. In addition to primary care advice, this finding also has the implication that other non-expert sources of estimates of risk such as web-based advice are likely to have serious limitations in their abilities to determine risk. Furthermore, as Collins (2000) proposes, this kind of tacit knowledge cannot be bottled and passed on in verbal descriptions, diagrams and instructions for action, but can only be learnt by extended experience observing and learning from the specialists themselves.

Transcription Notation

[Point of overlap onset
] Point of overlap termination
= Utterances follow each other rapidly
(0.0) Number in parentheses denotes duration of pause between utterances in seconds and tenths of seconds.
word Underlining indicates stress via pitch or amplitude
(( )) Double parentheses contain the researcher's observations or descriptions rather than what is heard.
Italics Indicates what is read rather than what is spoken

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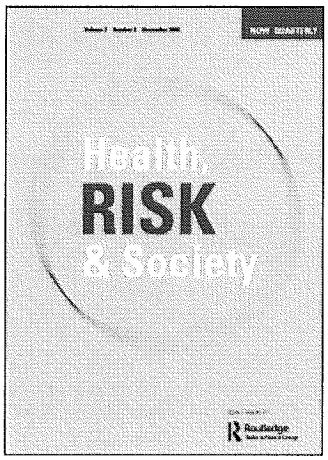

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Translations of risk: decision making in a cancer genetics service

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